Inqaba Biotechnical Industries (Pty) Ltd
Animal Genetics Division

Co. Reg. No: 2001/011245/07
VAT No: 4150197251

Congenital Myasthenic Syndrome (CMS) - Golden Retriever Type

Client Name:	Lauriaan Morton (LAU002)	Report No:	ZO2025/9673/20250804/#101088
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Profile:	DG2025/93667	Species:	Canis lupus familiaris / Canine / Dog
Name:	Golden Starseed Blue (Mufasa)	Microchip #:	972274200143122
Breed:	Golden Retriever		

Test:	[CMS_GR] Congenital Myasthenic Syndrome (CMS) - Golden Retriever Type		
Results:	c.880G>A	GG	Clear

Sample Type: Whole Blood (EDTA) Extraction Method: DNA Extraction: D4069 Test Type: Genetic Health

[CMS_GR] Congenital Myasthenic Syndrome (CMS) - Golden Retriever Type

Congenital Myasthenic Syndrome is a neuromuscular disorder, with clinical signs of the disease beginning at weaning, resulting in progressive muscle weakness exacerbated by exercise. The gait of affected dogs is often characterized by choppy, increasingly short strides, as well as stiffening of the legs.

The condition is caused by a c.880G>A substitution mutation in the collagen-like tail of the asymmetric acetylcholinesterase (COLQ) gene, which results in an amino acid change from glycine to arginine. This change inhibits normal gene function, and is suspected to affect the removal of acetylcholine from the neuromuscular junction, which is required for normal muscle function. The mutation is well characterised in the Golden Retriever.

The condition is inherited in an autosomal recessive manner, meaning that the dog must have two copies of the mutation to present with the condition.

References: Tsai et al 2019. Congenital Myasthenic Syndrome in Golden Retrievers is associated with a novel COLQ mutation J. Vet. Intern. Med. 2020 (34), 258-265.

Disclaimer:
Whatever is contained in this report is subject to the following:

This report does not disregard the existence of any rare or unknown variant within this gene or other gene(s) that may results in the same or similar trait. Multiple mutations and/or gene(s) may contribute to the overall trait observed.

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The DNA profile is based on a preliminary marker panel that is subject to modification pending additional genetic information.

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